illumina®

Discover the synergy of next-generation sequencing and arrays

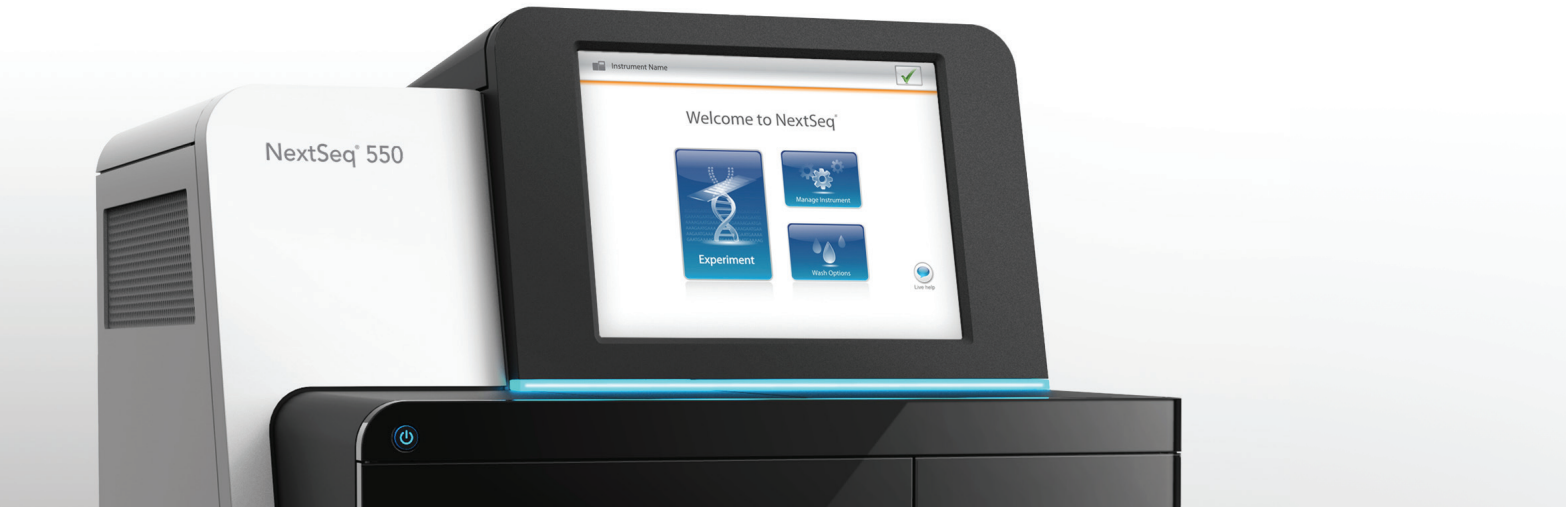
Combined testing for cytogenomic analysis using CytoSNP-850K+TruSight® One

Are you getting the full picture when it comes to clinical research? If you're conducting postnatal genetic investigations using only chromosomal arrays for copy number variants (CNV), you might be missing causal variants. To get a comprehensive view and broader scope of data analysis, labs are now adding sequencing for the detection of single nucleotide variants (SNV) that may be associated with genetic disease. The Infinium® CytoSNP-850K+TruSight One offers a comprehensive solution of arrays and sequencing—all on one platform—the NextSeq® 550.

The results you need. The confidence you want.

Combining the analysis from SNP arrays together with variant calls from sequencing, you'll gain confidence in your results. Leverage proven technologies that provide breadth across the genome for CNVs using microarrays and depth in the exonic regions of disease-causing genes with sequencing for SNVs.

The CytoSNP-850K BeadChip and TruSight One sequencing panels provide comprehensive analyses of genes associated with a clinical phenotype to respectively uncover copy number changes and sequence mutations with known consequence. The genomic targets of the TruSight One series of panels were curated by experts based on information in the Online Mendelian Inheritance in Man (OMIM®) and Human Gene Mutation Database (HGMD®). With TruSight One or TruSight One Expanded, consolidate and simplify your research portfolio with one sequencing assay designed to cover the most commonly ordered disease gene panels.



For Research Use Only. Not for use in diagnostic procedures.

CytoSNP-850K

- 18kb resolution for CNVs across the genome
- Dosage determination of >950 genes identified by the Deciphering Developmental Disorders (DDD) study and >3,100 OMIM morbid genes

TruSight One

- Targeted sequencing for >62,000 exons for >4,800 genes
- 12Mb sequencing of human genome
- Minimum 20× coverage of >95% of target regions

TruSight One Expanded

- Targeted sequencing for >86,000 exons for >6,700 genes
- 16.5Mb sequencing of human genome
- Minimum 20× coverage of >95% of target regions

One platform. A comprehensive solution.

The NextSeq 550 gives you the spectrum of data you want, and with intuitive bioinformatics tools, you can gain valuable insights on postnatal samples. Independent analyses for CNV and SNV can be profoundly impactful, but a centralized analysis pipeline that visualizes both CMA and NGS together can facilitate the discovery of heterogenic events for a more complete analysis. BioDiscovery's NxClinical™ v3.0 visualization tool accepts both array and sequencing data simultaneously in the same database to facilitate correlation of results. NxClinical™ v3.0 enables the interpretation of copy number, absence of heterozygosity (AOH), and nucleotide sequence variant changes together in one system, enhancing the cohesiveness of the dual analysis strategy.

Not just efficient, effective.

Costing less than whole genome sequencing and whole exome sequencing, the CytoSNP-850K+TruSight One analysis approach on the NextSeq 550 delivers optimum value—giving you the ability to maximize your investment with more testing ability on a single instrument. Plus, with the ability to observe genome-wide CNVs and SNVs in relevant genes, you'll be able to increase detection rates.

To learn more about our solutions in genetic health, visit www.illumina.com/GeneticHealth

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